

What Are Heritable Disorders of Connective Tissue?

Fast Facts: An Easy-to-Read Series of Publications for the Public

More than 200 heritable disorders of connective tissue (HDCTs) affect the tissues between the cells of your body. The disorders are called “heritable,” because they are passed on from parent to child. HDCTs come from changes to genes that build tissues.

Some HDCTs change the look and growth of skin, bones, joints, heart, blood vessels, lungs, eyes, and ears. Others change how these tissues work. Many, but not all, HDCTs are rare.

What Are Genes?

Genes carry our hereditary (family) information. We each have two copies of most genes: one set from each parent. Genes are what make you look like your biological family.

What Is Connective Tissue?

Connective tissue supports many parts of the body (skin, eyes, heart, etc.). Think of it as “cellular glue” that:

- Helps bring nutrients to the tissue
- Gives tissue form and strength
- Helps some of the tissues do their work.

Connective tissue is made of many kinds of proteins. Sometimes genes that have changed make proteins that don’t do their job right. This can change how the connective tissues work. Sometimes this leads to an HDCT.

What Are Some Kinds of Heritable Disorders of Connective Tissue?

Common HDCTs include:

- Ehlers-Danlos syndrome (EDS). This group of HDCTs mostly affects the skin and joints. With EDS, connective tissue becomes weak. This can cause fragile, sagging skin, and loose joints.
- Epidermolysis bullosa (EB). With these disorders, the skin blisters when it is stressed. For example, a hug could cause a blister.
- Marfan syndrome. This disorder can affect the heart, blood vessels, lungs, eyes, bones, and ligaments. People with this syndrome may be unusually tall and thin, with long arms and legs.
- Osteogenesis imperfecta (OI). With this disorder, bones break easily. Sometimes they break for no obvious reason.

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Who Gets Heritable Disorders of Connective Tissue?

There could be as many as 1 million people in the United States with an HDCT. It can affect anyone. Some of these disorders are obvious at birth. Others don't become obvious until later in life.

Does Anything Increase the Chances of Having a Genetic Disease?

Several things make people more likely to get or pass on a genetic disease:

- Parents who have a genetic disease
- A family history of a genetic disease
- Parents who are closely related
- Parents who come from a region where the defect is common
- Parents who don't have disease symptoms but "carry" a certain gene (sometimes this gene is found through genetic testing).

What Are the Symptoms of Heritable Disorders of Connective Tissue?

Each HDCT has its own symptoms. Some examples are:

- Bone growth problems. People with bone growth disorders can have brittle bones. They can also have bones that are too long or too short.
- Joint issues. Some HDCTs cause joints to be too loose or too tight.
- Skin problems. There are HDCTs that cause loose skin, skin that hangs in folds, or blistered skin.
- Blood vessel damage. Some HDCTs lead to weak blood vessels. Other HDCTs can close-off or block blood vessels.
- Height issues. Some HDCTs cause people to be unusually tall or short.
- Head and facial structural problems. Certain HDCTs can make the head and face look different from others.

How Do Doctors Diagnose Heritable Disorders of Connective Tissue?

To diagnose HDCTs, doctors look at:

- Family history
- Medical history
- Results from a physical exam.

Some people may also see a medical geneticist (someone who studies how genes affect people). Lab tests can confirm some HDCTs, but not all.

What Treatments Are Available?

There are certain ways to manage and treat each disorder. But in general, people with HDCTs should:

- Take care of their health
- Stay in touch with doctors who will know about new treatments
- Have regular checkups so doctors can check for changes or problems.

What Research Is Being Done on Heritable Disorders of Connective Tissue?

Experts are trying to:

- Figure out where changes (mutations) are in the connective tissue genes
- Find out which changes cause the HDCTs
- Try to find out how these changes cause the HDCT
- Use all the new knowledge to plan and test new kinds of therapy.

Other research looks at:

- Ways to use gene therapy
- Gene changes that cause bone disease
- Groups of proteins that cause tissue to be stiff
- Cells that form the body's tissues
- Aneurysms (weak spots in blood vessel walls that can burst)
- Drugs that can be used to treat brittle bones
- Mind-body therapy for chronic pain
- Bone growth that isn't normal
- Gene defects that cause elastin to not work right (elastin is what lets tissues stretch).

For More Information on Heritable Disorders of Connective Tissue and Other Related Conditions:

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The information in this publication was summarized in easy-to-read format from information in a more detailed NIAMS publication. To order the Heritable Disorders of the Connective Tissue Q&A full-text version, please contact NIAMS using the contact information above. To view the complete text or to order online, visit <http://www.niams.nih.gov>.